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What is claimed is:

CLAIMS

1. A method for screening for the presence of a PXE mutation, the method comprising the steps of:

interrogating an MRP6 nucleic acid in a patient sample for the presence of a PXE mutation; and,

identifying a positive screen as the presence of said mutation.

2. The method according to claim 1, wherein the said patient sample is selected from the group consisting of blood, saliva, amniotic fluid, and tissue.

3. The method according to claim 2, wherein the said patient sample is blood.

4. The method according to claim 1, wherein said interrogating step is a nucleic acid sequence scanning assay.

5. The method according to claim 4, wherein said scanning assay is selected from the group consisting of SSCP, DGGE, RFLP, LCR, DHPLC, and enzymatic cleavage.

6. The method according to claim 1, wherein said interrogating step is a specific mutation detection assay.

7. The method according to claim 6, wherein said detection assay is selected from the group consisting of oligonucleotide hybridization and primer extension assays.

8. The method according to claim 1, wherein said interrogating step is a nucleic acid sequencing assay.

9. The method according to claim 1, wherein said assay detects the presence of a mutation selected from the group consisting of a deletion, a substitution, an insertion, and a rearrangement.

10. The method according to claim 1, wherein said mutation is a mutation in codon 1141.

11. The method according to claim 1, wherein said mutation is a deletion of base 3775.

12. The method according to claim 1, wherein said mutation is a non-conserved amino acid substitution.

13. The method according to claim 1, wherein said mutation is in a codon selected from the group consisting of 1114, 1138, 1141, 1298, 1302, 1303, 1314, and 1321.

14. The method according to claim 1, wherein said mutation is in a splice site in an intron.

15. The method according to claim 1, wherein said mutation is in the promoter region of the MRP6 gene.

16. The method according to claim 1, wherein said mutation is in a polyA site of the MRP6 gene

17. The method according to claim 1, wherein said nucleic acid is selected from the group consisting of mRNA, genomic DNA, and cDNA.

18. A method for screening for the presence of a PXE mutation, the method comprising the steps of:

detecting MRP6 expression; and,

identifying a positive screen as one that detects a level of MRP6 expression that is lower than normal MRP6 expression.

19. The method of claim 18, wherein said detecting step detects MRP6 mRNA.

20. The method of claim 18, wherein said detecting step detects MRP6 protein.

21. The method according to claim 20, wherein said detecting step is an antibody-based assay.

22. The method according to claim 21, wherein said assay is selected from the group consisting of an ELISA and a Western blot.

23. A method for screening for the presence of a PXE mutation, the method comprising the steps of:

interrogating an MRP6 protein in a patient for the presence of a PXE protein change; and,

identifying a positive screen as the presence of said change.

24. The method according to claim 23, wherein said interrogating step measures the size of the MRP6 protein.

25. The method according to claim 1, wherein said assay detects the presence of a mutation in an exon of the MRP6 gene.

26. The method according to claim 25, wherein said exon is selected from exons 1-31 of the MRP6 gene

27. The method according to claim 1, wherein said interrogating step is a hybridization assay.

28. An oligonucleotide for use in an assay to detect a PXE associated mutation, wherein said oligonucleotide hybridizes to a PXE mutation in an MRP6 nucleic acid under stringent conditions.

29. An oligonucleotide of claim 32, wherein said oligonucleotide is selected from the group consisting of the oligonucleotides of SEQ ID NO: 10 through SEQ ID NO: 25.

30. A method for identifying a patient at risk of having children with PXE, the method comprising the step of interrogating a patient sample for the presence of a PXE mutant MRP6 allele.

31. A method for identifying a patient at risk of developing a PXE associated symptom, the method comprising the step of interrogating a patient sample for the presence of a PXE mutant MRP6 allele.

32. A method for diagnosing PXE in a patient, the method comprising the step of interrogating a patient sample for the presence of two PXE mutant MRP6 alleles.

33. The method of claim 32, wherein said patient is a homozygous PXE patient.